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Sirenomelia - A Case Report

Case capsule

A 28-year-old third gravida with two previous normal deliveries and healthy children was admitted in Government district headquarters hospital, Kumbakonam with labor pain. She was a non consanguineous mother without any antenatal history of exanthematous fever and drugs. She was unbooked and had no proper screening ultrasound examination. She delivered per vaginally an alive male child with fused lower limbs. The baby cried well after birth with an Apgar of 9/10. The other physical findings (Figure 1) were

1. Imperforate anus
2. Fused genitalia with rudimentary penis
3. Single umbilical artery
4. The rotation of knees and feet was reversed with sole and the popliteal region facing forward.

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Sirenomelia is a congenital structural anomaly characterized by abnormal development of the caudal region of the body with varying degrees of fusion of lower limbs.

There were no obvious anomalies in the skull, facies, chest and lumbar region. The baby passed urine through the rudimentary penis. A provisional diagnosis of sirenomelia was made and further work up was planned. The mother absconded with the baby denying further investigations due to cultural constraints. Repeated postal communications evoked no response but the baby died in its first week of life.

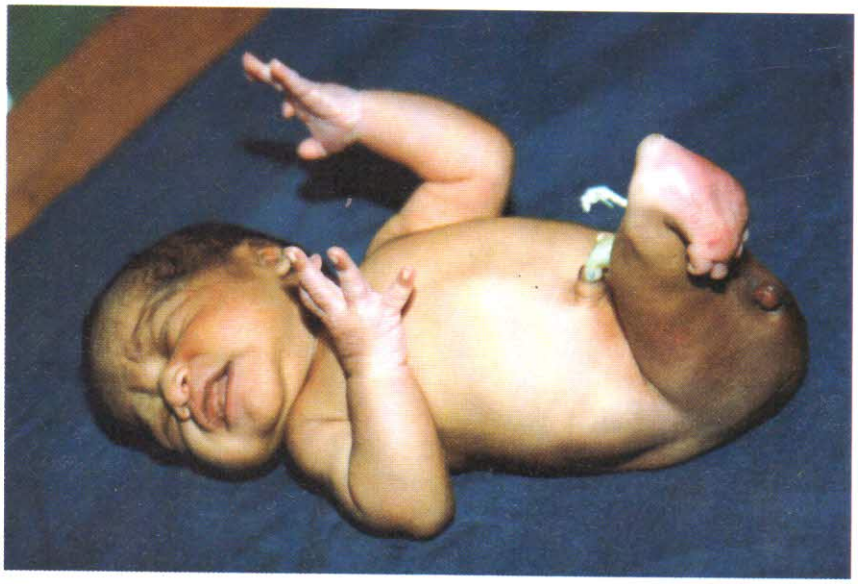

Figure 1. Case of sirenomelia.

Table 1
Classification of sirenomelia

		Femur	Tibia	Feet
A	Simpus apus	one	one	nil
B	Simpus unipus	two	two	one
C	Simpus dipus	one	one	two

Case discussion

Sirenomelia is a congenital structural anomaly characterized by abnormal development of the caudal region of the body with varying degrees of fusion of lower limbs[1]. It bears the resemblance of Mermaid of Greek mythology and hence the synonym Mermaid syndrome[2]. The prevalence of this anomaly is 1/100,000 live births with a male:female ratio of 3:1. About 300 cases are reported with eight from India[3]. Sirenomelia is simply classified into three types (Table 1).

The other classification is as shown in figure 2.

Etiology

The precise etiology is of sirenomelia not known but various theories have been proposed.

1. Intravenous administration of cadmium[4].
2. Antenatal administration and effects of vitamin A[5].
3. It can be considered as a manifestation of Caudal regression syndrome which itself is a consequence of abnormal development of structures derived from caudal mesoderm of the embryo before the fourth week of gestation[6].
4. Many authors have drawn the attention of overlap of phenotypic manifestation of VATER anomalies and sirenomelia[7].
5. Stevenson proposed vascular steal theory[8] in which a large artery arose from the aorta high in the abdomen beyond which aorta and its branches are hypoplastic.
6. Adhesions of amniotic bands[9] disrupted the fetal parts causing multiple anomalies.

The manifestations (cardinal features)[10] associated with sirenomelia are:

- Urogenital anomalies – Frequent bilateral renal agenesis, absent bladder simplified to no external genitals.
- Imperforate anus.
- Vertebral column anomalies like Sacral and coccygeal agenesis and disorganization of lumbar vertebrae including spina bifida.
- Radial aplasia, tracheo-esophageal fistula, cardiopathy cleft lip and palate are also reported.
- Single umbilical artery.
- The rotation of knees and feet is reversed with sole and the popliteal region facing forward.

The genetic basis is thought to be sporadic and nonrecurring. It's ideal to have antenatal screening and advise medical termination if diagnosed. Usually they die in a

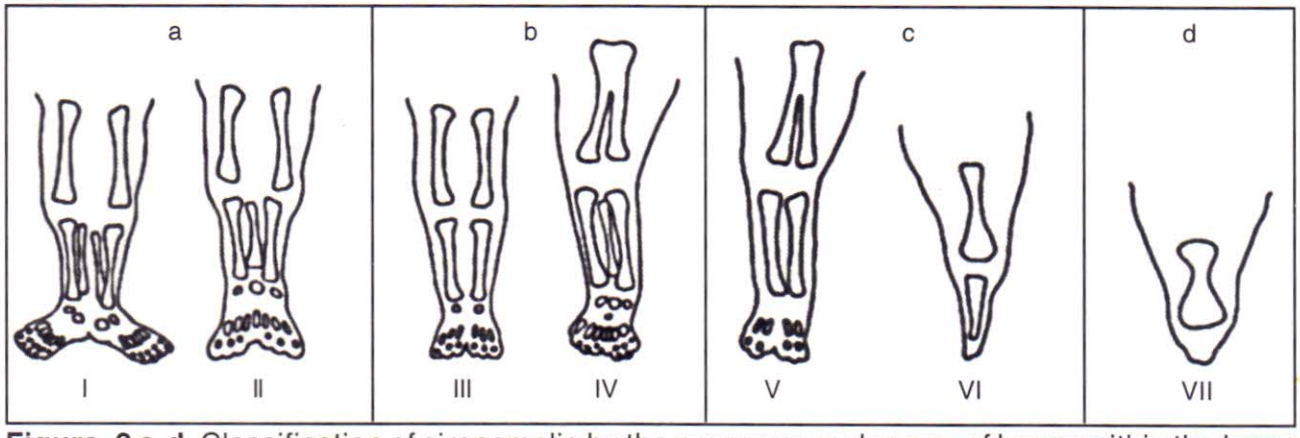

Figure. 2 a-d. Classification of sirenomelia by the presence or absence of bones within the lower limb. Type I = all bones of thigh and lower leg present; type II = fused fibula; type III = fibula absent; type IV = partially fused femur, fused fibula; type V = Partially fused femur; type VI = fused femur, fused tibia; type VII = fused femur, tibia absent.

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day or two after birth. Only a handful without renal and intestinal anomalies has survived longer after major reconstructive surgeries[11]. In our case we could find many of the features. Unfortunately we could not further investigate to clinch the etiology or establish the other features due to non cooperation from the parent side. Still we opted to present the case for its rarity.

Acknowledgements

We would like to thank Dr S. Rajendran, Superintendent, Govt. Dist. Headquarters Hospital Kumbakonam for permitting to publish the article.

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